

Validation of Forensic Identity Tests: Concordance Study with MiniFiler and other STR Kits

NIST has conducted studies to help ensure the accuracy of new forensic identity tests specifically designed for DNA samples of poor quality as a result of fire or other disaster. These new genetic tests analyze very short sequences of DNA called miniature short tandem repeats (miniSTR.) These assays were demonstrated during efforts to identify the 9/11 World Trade Center victims. The first commercial kit based on this miniSTR technology is now being tested and promises to greatly aid forensic labs in the future to solve previously intractable cases due to poor DNA sample quality.

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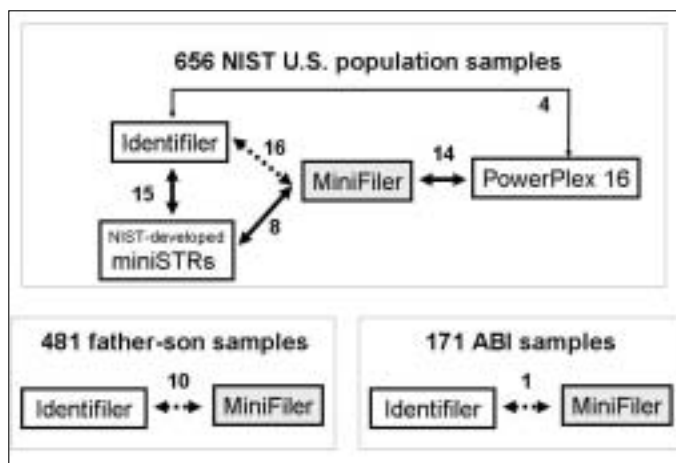
A number of studies have demonstrated that successful analysis of degraded DNA specimens from mass disasters or forensic evidence improves with smaller sized polymerase chain reaction (PCR) products. If DNA is exposed to the elements or to fire for any length of time, degradation can occur due to bacterial, biochemical or oxidative processes. Within the forensic community, a core set of short tandem repeat (STR) markers have been developed for utilization in forensic casework, and large DNA databases such as the Combined DNA Index System (CODIS) have been developed incorporating these markers.

NIST scientists are assisting law enforcement by helping facilitate the development of DNA-based human identity tests to help solve crimes unable to be prosecuted because of poor DNA quality samples.

During the spring and summer of 2006, the NIST Human Identity Project Team was involved in beta-testing a new DNA testing kit that will enable improved recovery of genetic information from biological samples that are highly fragmented or otherwise environmentally compromised. The principles behind miniSTR technology were pioneered at NIST in order to aid recovery of information from the highly fragmented remains of victims from the 9/11/01 terrorist attacks on the World Trade Center twin towers.

Applied Biosystems (Foster City, CA) is working with NIST scientists to validate their AmpF ℓ STR[®] MiniFiler[™] PCR Amplification kit through examining samples

amplified with this new test and presently available DNA tests. These concordance tests are necessary because each test contains slightly different PCR primers that target the same regions of human DNA. Due to the fact that sequence variation can and does exist in the flanking region of STR markers, samples need to be evaluated to verify PCR amplification performance and quantify any differences where mutations are occurring in primer binding regions.



The figure illustrates the differences noted between the various tested STR kits during concordance studies conducted.

The MiniFiler kit enables size reduction on eight of the larger STR loci amplified in the other commercial STR kits such as Identifiler (Applied Biosystems) and PowerPlex 16 (Promega Corporation, Madison, WI). MiniFiler amplifies CSF1PO, FGA, D2S1338, D7S820, D13S317, D16S539, D18S51, and D21S11 as well as the sex-typing locus amelogenin. A total of 1,308 samples were evaluated with both the MiniFiler and Identifiler STR kits: 449 African American, 445 Caucasian, 207 Hispanic, and 207 Asian individuals. Full concordance between Identifiler and MiniFiler kits was observed in 99.7% (10,437 out of 10,464) STR allele calls compared. The 27 differences observed (see dotted lines in figure) encompass the loci D13S317 (n=14) and D16S539 (n=10) as well as D18S51 (n=1), D7S820 (n=1), and CSF1PO (n=1). Genotyping discrepancies between the Identifiler and MiniFiler kits were confirmed by re-amplification of the samples and further testing using the PowerPlex 16 kit. DNA sequence analysis was also performed in order to understand the nature of the genetic variations causing the allele dropout or apparent repeat unit shift.

Publications:

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